



7. Factor V deficiency

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Patient comment 1: "I told the ER doc I had factor V Leiden, but he kept calling it factor V deficiency, and he thought it caused irregular bleeding instead of clotting."

Factor V Leiden and factor V deficiency are two completely separate problems. Unfortunately, they are sometimes confused by physicians, who do not know what factor V Leiden is.

Patient comment 2: "When my mother and father went to be tested the doctor tested them for factor V activity instead of the genetic test."

Either the physician ordered the wrong test, or the physician ordered the right test but the person entering the order into the computer entered the wrong test. It is not helpful to check factor V activity levels in patients with factor V Leiden, because they are normal.

Patient comment 3: "Somebody please correct me if I am wrong: there are not two different types of factor V!!!"

There is only one type of normal factor V. And there is only one type of mutated factor V Leiden (also termed (R506Q or Arg506Gln; also referred to as G1691A factor V gene mutation). You can have 1 bad gene for factor V Leiden (= heterozygous) or 2 bad genes (= homozygous). There are many different types of mutated factor Vs that cause factor V deficiency.

Patient comment 4: "I am very interested in the 2 different FVL."

As mentioned above, there is only one FVL (= factor V Leiden).

Patient comment 5: "What is factor V Leiden deficiency?"

That does not exist. However, it is terminology that is not infrequently, though incorrectly, used by physicians. Typically the physicians mean "factor V Leiden".

Factor V deficiency is an inherited disorder, in which the clotting factor V is low. A bleeding problem results. The disorder is very rare, occurring in only one in 1 million people. Thus, there are only approximately 270 patients in the United States with this disorder. It is inherited as an autosomal recessive disorder, which means that an individual needs to be homozygous for the disorder (i.e. have 2 bad genes) to develop symptoms. Patients may have bleeding as newborns. However, 50 % of patients are not diagnosed until adulthood. The diagnosis is made by finding low factor V activity levels, usually between 1 and 20 % of normal. If treatment is needed, fresh frozen plasma (= FFP) is typically given.

Factor V Leiden is a completely different inherited disorder, in which factor V is mutated in a specific spot (R506Q, also termed Arg506Gln; also referred to as factor V gene mutation G1691A), leading to a tendency to clot too easily. The mutation is very common, occurring in 5% of the U.S. American population. A few calculations:

- * 1 in 20 U.S. Americans are heterozygous (= have 1 bad gene) for FVLeiden, i.e. 13.5 million people in the U.S.;
- * 1 in 1,600 U.S. Americans are homozygous (= have 2 bad genes) for FVLeiden, i.e. 170,000 people in the U.S.;
- * factor V Leiden is thus fifty thousand times more common than factor V deficiency.

Factor V activity levels in patients with factor V Leiden are normal. It is therefore not useful to obtain them. The diagnosis of FVLeiden is made by genetic testing, called PCR (= polymerase chain reaction) or, less precisely, by APC-resistance assay (= activated protein C resistance assay). If treatment is needed, it consists of blood thinning medications.